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Case Report Section

t(6;17)(p21;p13) associated with t(3;3)(q21;q26.2) in AML

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Abstract

Case report on t(6;17)(p21;p13) associated with t(3;3)(q21;q26.2) in AML.

Clinics

Age and sex
25 years old female patient.

Previous history
Preleukemia; no previous malignancy, no inborn condition of note

Organomegaly
No hepatomegaly , no splenomegaly , no enlarged lymph nodes , no central nervous system involvement

Blood

WBC: 20X 10^9/l
HB: 86g/dl
Platelets: 14X 10^9/l
Blasts: 12%
Bone marrow: Hyperplastic bone marrow with dyserythropoiesis, megaloblastic erythropoiesis, presence of multinuclear forms, depressed megakaryocytes and 23% blasts.

Cyto-Pathology Classification

Phenotype: Acute myeloid leukemia

Immunophenotype
Positive for CD13, CD33, CD34, HLDR, CS45, CD11b, MPO and CD64, dim expression of CD14 and CD117.

Diagnosis
AML with t(3;3)(q21;q26.2)

Survival

Date of diagnosis: 12-2017
Complete remission: no
Last follow up: 12-2017

Karyotype

Sample bone marrow

Culture time 24h

Banding G-banding

Results
45,XX,t(3;3)(q21;q26.2),t(6;17)(p21;p13),-7[14]/45,XY,t(3;3)(q21;q26.2),-7[4]/ 46,XX [2]

Figure 1. Karyotype of the patient with t(3;3)(q21;q26.2) and -7 (A) and with t(6;17)(p21;p13) as an additional anomaly (B)

Other molecular cytogenetics technics

Fluorescence in situ hybridization studies (FISH) were performed using Kreatechô MECOM (EVI1) t(3;3); inv(3)(3q26) Break FISH probe and SureFISH 17p13.3 (PAFAH1B1) and RUNX2 (6p21.1) probes.

Other molecular cytogenetics results

Hybridization with EVI1 probe confirmed the rearrangement of the gene as a result of t(3;3)(q21;q36.2) on 10 metaphases and in 90 % of interphase cells. Hybridization with Vysis D7S486/ Vysis CEP 7 probe showed monosomy 7 in 90% of cells. FISH studies with SureFISH PAFAH1B1 and RUNX2 probes showed normal signals on 2 metaphases without t(6;17) and juxtaposition of PAFAH1B1 from 17p13.3 to 6p21.1 at the site of RUNX2 gene in 5 metaphases.

Comments

We described here an AML patient with t(3;3)(q21;q26.2) associated with monosomy 7 and a rare translocation t(6;17)(p21;p13) (La Starza et al., 26). The chromosomal t(6;17)(p21;p13) was found in a sideline as an additional anomaly to t(3;3)(q21;q26.2) and monosomy 7, therefore likely representing a secondary aberration to these primary anomalies. inv(3)(q21q26.2) or t(3;3)(q21;q26.2) in AML or MDS result in deregulation of the proto-oncogene EVI1 (MECOM), which affect the RAS/receptor tyrosine kinase pathway. The concomitant monosomy 7 and t(6;17)(p21;p13) are probably cooperating genetic lesions that developed during malignant transformation processes in our patient.

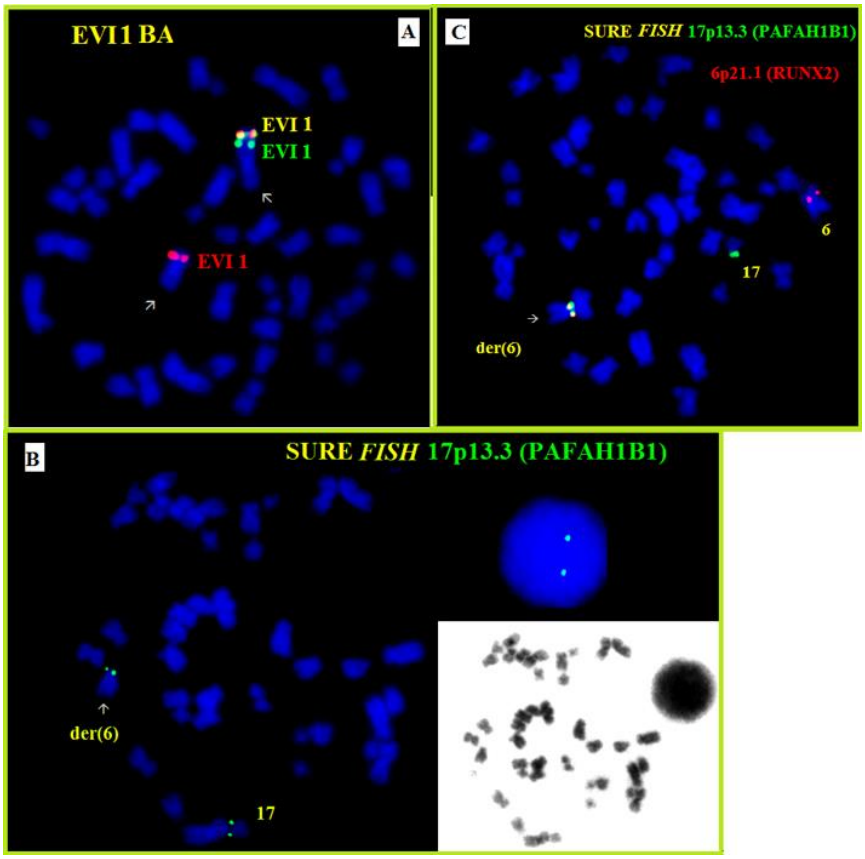

Figure 2. (A) Fluorescence in situ hybridization with Kreatechô MECOM (EVI1) break apart probe confirmed the rearrangement of the gene as a result of t(3;3)(q21;q36.2). FISH studies with SureFISH PAFAH1B1 located at 17p.13.3 revealed juxtaposition of PAFAH1B1 from 17p13.3 to der(6) chromosome (B). Simultaneous hybridization with SureFISH PAFAH1B1 and RUNX2 probes showed cohybridization of PAFAH1B1 and RUNX2 genes on 6p21.1 (C).

References

La Starza R, Aventin A, Matteucci C, Crescenzi B, Romoli S, Testoni N, Pierini V, Ciolli S, Sambani C, Locasciulli A, Di Bona E, Lafage-Pochitaloff M, Martelli MF, Marynen P, Mecucci C. Genomic gain at 6p21: a new cryptic molecular rearrangement in secondary myelodysplastic syndrome and acute myeloid leukemia. Leukemia. 2006 Jun;20(6):958-64

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